

SEQUENCE LISTING

<110> Keating, Mark T.
Splawski, Igor

<120> MUTATIONS IN AND GENOMIC STRUCTURE OF HERG - A LONG QT
SYNDROME GENE

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<150> 09/122,847

<151> 1998-07-27

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<170> PatentIn Ver. 2.0

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 Gln Ile Thr Leu Asn Phe Val Asp Leu Lys Gly Asp Pro Phe Leu Ala
 335 340 345 350

tcg ccc acc agt gac cgt gag atc ata gca cct aag ata aag gag cga 1164
 Ser Pro Thr Ser Asp Arg Glu Ile Ile Ala Pro Lys Ile Lys Glu Arg
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acc cac aat gtc act gag aag gtc acc cag gtc ctg tcc ctg ggc gcc 1212
 Thr His Asn Val Thr Glu Lys Val Thr Gln Val Leu Ser Leu Gly Ala
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gac gtg ctg cct gag tac aag ctg cag gca ccg cgc atc cac cgc tgg 1260
 Asp Val Leu Pro Glu Tyr Lys Leu Gln Ala Pro Arg Ile His Arg Trp
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acc atc ctg cat tac agc ccc ttc aag gcc gtg tgg gac tgg ctc atc 1308
 Thr Ile Leu His Tyr Ser Pro Phe Lys Ala Val Thr Asp Trp Leu Ile
 400 405 410

ctg ctg ctg gtc atc tac acg gct gtc ttc aca ccc tac tcg gct gcc 1356
 Leu Leu Leu Val Ile Tyr Thr Ala Val Phe Thr Pro Tyr Ser Ala Ala
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ttc ctg ctg aag gag acg gaa gaa ggc ccg cct gct acc gag tgt ggc 1404
 Phe Leu Leu Lys Glu Thr Glu Glu Gly Pro Pro Ala Thr Glu Cys Gly
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 Tyr Ala Cys Gln Pro Leu Ala Val Val Asp Leu Ile Val Asp Ile Met
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 Phe Ile Val Asp Ile Leu Ile Asn Phe Arg Thr Thr Tyr Val Asn Ala
 465 470 475

aac gag gag gtg gtc agc cac ccc ggc cgc atc gcc gtc cac tac ttc 1548
 Asn Glu Glu Val Val Ser His Pro Gly Arg Ile Ala Val His Tyr Phe
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09735505 4400

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gcg cgg ctg ctg cgg ctg gtg cgc gtg gcg cgg aag ctg gat cgc tac 1692
 Ala Arg Leu Leu Arg Leu Val Arg Val Ala Arg Lys Leu Asp Arg Tyr 540
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 Ser Glu Tyr Gly Ala Ala Val Leu Phe Leu Leu Met Cys Thr Phe Ala 555
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 Leu Ile Ala His Trp Leu Ala Cys Ile Trp Tyr Ala Ile Gly Asn Met 570
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gag cag cca cac atg gac tca cgc atc ggc tgg ctg cac aac ctg ggc 1836
 Glu Gln Pro His Met Asp Ser Arg Ile Gly Trp Leu His Asn Leu Gly 590
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 Asp Gln Ile Gly Lys Pro Tyr Asn Ser Ser Gly Leu Gly Gly Pro Ser 605
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 Ile Lys Asp Lys Tyr Val Thr Ala Leu Tyr Phe Thr Phe Ser Ser Leu 620
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acc agt gtg ggc ttc ggc aac gtc tct ccc aac acc aac tca gag aag 1980
 Thr Ser Val Gly Phe Gly Asn Val Ser Pro Asn Thr Asn Ser Glu Lys 635
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 Ile Phe Gly Asn Val Ser Ala Ile Ile Gln Arg Leu Tyr Ser Gly Thr 670
 655 660 665

gcc cgc tac cac aca cag atg ctg cgg gtg cgg gag ttc atc cgc ttc 2124
 Ala Arg Tyr His Thr Gln Met Leu Arg Val Arg Glu Phe Ile Arg Phe 685
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 Gly Phe Pro Glu Cys Leu Gln Ala Asp Ile Cys Leu His Leu Asn Arg 730
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tca ctg ctg cag cac tgc aaa ccc ttc cga ggg gcc acc aag ggc tgc 2316
 Ser Leu Leu Gln His Cys Lys Pro Phe Arg Gly Ala Thr Lys Gly Cys 750
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 Leu Arg Ala Leu Ala Met Lys Phe Lys Thr Thr His Ala Pro Pro Gly
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gac aca ctg gtg cat gct ggg gac ctg ctc acc gcc ctg tac ttc atc 2412
 Asp Thr Leu Val His Ala Gly Asp Leu Leu Thr Ala Leu Tyr Phe Ile
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ctg ggg aag aat gac atc ttt ggg gag cct ctg aac ctg tat gca agg 2508
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cct ggc aag tcg aac ggg gat gtg cgg gcc ctc acc tac tgt gac cta 2556
 Pro Gly Lys Ser Asn Gly Asp Val Arg Ala Leu Thr Tyr Cys Asp Leu
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 His Lys Ile His Arg Asp Asp Leu Leu Glu Val Leu Asp Met Tyr Pro
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 Glu Phe Ser Asp His Phe Trp Ser Ser Leu Glu Ile Thr Phe Asn Leu
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 Arg Asp Thr Asn Met Ile Pro Gly Ser Pro Gly Ser Thr Glu Leu Glu
 865 870 875

ggt ggc ttc agt cgg caa cgc aag cgc aag ttg tcc ttc cgc agg cgc 2748
 Gly Gly Phe Ser Arg Gln Arg Lys Arg Lys Leu Ser Phe Arg Arg Arg
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 Thr Asp Lys Asp Thr Glu Gln Pro Gly Glu Val Ser Ala Leu Gly Pro
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gat gag ggc cca ggc cgc agc tcc agc ccc ctc cgc ctg gtg ccc ttc 2940
 Asp Glu Gly Pro Gly Arg Ser Ser Ser Pro Leu Arg Leu Val Pro Phe
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 Ser Ser Pro Arg Pro Pro Gly Glu Pro Pro Gly Gly Glu Pro Leu Met
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 Glu Asp Cys Glu Lys Ser Ser Asp Thr Cys Asn Pro Leu Ser Gly Ala
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 Phe Ser Gly Val Ser Asn Ile Phe Ser Phe Trp Gly Asp Ser Arg Gly
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 Arg Gln Tyr Gln Glu Leu Pro Arg Cys Pro Ala Pro Thr Pro Ser Leu
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 Leu Asn Ile Pro Leu Ser Ser Pro Gly Arg Arg Pro Arg Gly Asp Val
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 Glu Ser Arg Leu Asp Ala Leu Gln Arg Gln Leu Asn Arg Leu Glu Thr
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 atg acg ctg gtc ccg ccc gcc tac agt gct gtg acc acc ccg ggg cct 3324
 Met Thr Leu Val Pro Pro Ala Tyr Ser Ala Val Thr Thr Pro Gly Pro
 1075 1080 1085
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 Gly Pro Thr Ser Thr Ser Pro Leu Leu Pro Val Ser Pro Leu Pro Thr
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 Glu Leu Pro Pro Gly Ala Pro Glu Leu Pro Gln Glu Gly Pro Thr Arg
 1120 1125 1130
 cgc ctc tcc cta ccg ggc cag ctg ggg gcc ctc acc tcc cag ccc ctg 3516
 Arg Leu Ser Leu Pro Gly Gln Leu Gly Ala Leu Thr Ser Gln Pro Leu
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 cac aga cac ggc tcg gac ccg ggc agt tagtggggct gccagtggtg 3563
 His Arg His Gly Ser Asp Pro Gly Ser
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 35 40 45
 Cys Glu Leu Cys Gly Tyr Ser Arg Ala Glu Val Met Gln Arg Pro Cys
 50 55 60
 Thr Cys Asp Phe Leu His Gly Pro Arg Thr Gln Arg Arg Ala Ala Ala
 65 70 75 80
 Gln Ile Ala Gln Ala Leu Leu Gly Ala Glu Glu Arg Lys Val Glu Ile
 85 90 95
 Ala Phe Tyr Arg Lys Asp Gly Ser Cys Phe Leu Cys Leu Val Asp Val
 100 105 110
 Val Pro Val Lys Asn Glu Asp Gly Ala Val Ile Met Phe Ile Leu Asn
 115 120 125
 Phe Glu Val Val Met Glu Lys Asp Met Val Gly Ser Pro Ala His Asp
 130 135 140
 Thr Asn His Arg Gly Pro Pro Thr Ser Trp Leu Ala Pro Gly Arg Ala
 145 150 155 160
 Lys Thr Phe Arg Leu Lys Leu Pro Ala Leu Leu Ala Leu Thr Ala Arg
 165 170 175
 Glu Ser Ser Val Arg Ser Gly Gly Ala Gly Gly Ala Gly Ala Pro Gly
 180 185 190
 Ala Val Val Val Asp Val Asp Leu Thr Pro Ala Ala Pro Ser Ser Glu
 195 200 205
 Ser Leu Ala Leu Asp Glu Val Thr Ala Met Asp Asn His Val Ala Gly
 210 215 220
 Leu Gly Pro Ala Glu Glu Arg Arg Ala Leu Val Gly Pro Gly Ser Pro
 225 230 235 240
 Pro Arg Ser Ala Pro Gly Gln Leu Pro Ser Pro Arg Ala His Ser Leu
 245 250 255
 Asn Pro Asp Ala Ser Gly Ser Ser Cys Ser Leu Ala Arg Thr Arg Ser
 260 265 270
 Arg Glu Ser Cys Ala Ser Val Arg Arg Ala Ser Ser Ala Asp Asp Ile
 275 280 285
 Glu Ala Met Arg Ala Gly Val Leu Pro Pro Pro Pro Arg His Ala Ser
 290 295 300

Thr Gly Ala Met His Pro Leu Arg Ser Gly Leu Leu Asn Ser Thr Ser
 305 310 315 320
 Asp Ser Asp Leu Val Arg Tyr Arg Thr Ile Ser Lys Ile Pro Gln Ile
 325 330 335
 Thr Leu Asn Phe Val Asp Leu Lys Gly Asp Pro Phe Leu Ala Ser Pro
 340 345 350
 Thr Ser Asp Arg Glu Ile Ile Ala Pro Lys Ile Lys Glu Arg Thr His
 355 360 365
 Asn Val Thr Glu Lys Val Thr Gln Val Leu Ser Leu Gly Ala Asp Val
 370 375 380
 Leu Pro Glu Tyr Lys Leu Gln Ala Pro Arg Ile His Arg Trp Thr Ile
 385 390 395 400
 Leu His Tyr Ser Pro Phe Lys Ala Val Trp Asp Trp Leu Ile Leu Leu
 405 410 415
 Leu Val Ile Tyr Thr Ala Val Phe Thr Pro Tyr Ser Ala Ala Phe Leu
 420 425 430
 Leu Lys Glu Thr Glu Glu Gly Pro Pro Ala Thr Glu Cys Gly Tyr Ala
 435 440 445
 Cys Gln Pro Leu Ala Val Val Asp Leu Ile Val Asp Ile Met Phe Ile
 450 455 460
 Val Asp Ile Leu Ile Asn Phe Arg Thr Thr Tyr Val Asn Ala Asn Glu
 465 470 475 480
 Glu Val Val Ser His Pro Gly Arg Ile Ala Val His Tyr Phe Lys Gly
 485 490 495
 Trp Phe Leu Ile Asp Met Val Ala Ala Ile Pro Phe Asp Leu Leu Ile
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 Phe Gly Ser Gly Ser Glu Glu Leu Ile Gly Leu Leu Lys Thr Ala Arg
 515 520 525
 Leu Leu Arg Leu Val Arg Val Ala Arg Lys Leu Asp Arg Tyr Ser Glu
 530 535 540
 Tyr Gly Ala Ala Val Leu Phe Leu Leu Met Cys Thr Phe Ala Leu Ile
 545 550 555 560
 Ala His Trp Leu Ala Cys Ile Trp Tyr Ala Ile Gly Asn Met Glu Gln
 565 570 575
 Pro His Met Asp Ser Arg Ile Gly Trp Leu His Asn Leu Gly Asp Gln
 580 585 590
 Ile Gly Lys Pro Tyr Asn Ser Ser Gly Leu Gly Gly Pro Ser Ile Lys
 595 600 605
 Asp Lys Tyr Val Thr Ala Leu Tyr Phe Thr Phe Ser Ser Leu Thr Ser
 610 615 620
 Val Gly Phe Gly Asn Val Ser Pro Asn Thr Asn Ser Glu Lys Ile Phe
 625 630 635 640

Ser Ile Cys Val Met Leu Ile Gly Ser Leu Met Tyr Ala Ser Ile Phe
 645 650 655
 Gly Asn Val Ser Ala Ile Ile Gln Arg Leu Tyr Ser Gly Thr Ala Arg
 660 665 670
 Tyr His Thr Gln Met Leu Arg Val Arg Glu Phe Ile Arg Phe His Gln
 675 680 685
 Ile Pro Asn Pro Leu Arg Gln Arg Leu Glu Glu Tyr Phe Gln His Ala
 690 695 700
 Trp Ser Tyr Thr Asn Gly Ile Asp Met Asn Ala Val Leu Lys Gly Phe
 705 710 715 720
 Pro Glu Cys Leu Gln Ala Asp Ile Cys Leu His Leu Asn Arg Ser Leu
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 Leu Gln His Cys Lys Pro Phe Arg Gly Ala Thr Lys Gly Cys Leu Arg
 740 745 750
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 755 760 765
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 770 775 780
 Gly Ser Ile Glu Ile Leu Arg Gly Asp Val Val Val Ala Ile Leu Gly
 785 790 795 800
 Lys Asn Asp Ile Phe Gly Glu Pro Leu Asn Leu Tyr Ala Arg Pro Gly
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 Lys Ser Asn Gly Asp Val Arg Ala Leu Thr Tyr Cys Asp Leu His Lys
 820 825 830
 Ile His Arg Asp Asp Leu Leu Glu Val Leu Asp Met Tyr Pro Glu Phe
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 Ser Asp His Phe Trp Ser Ser Leu Glu Ile Thr Phe Asn Leu Arg Asp
 850 855 860
 Thr Asn Met Ile Pro Gly Ser Pro Gly Ser Thr Glu Leu Glu Gly Gly
 865 870 875 880
 Phe Ser Arg Gln Arg Lys Arg Lys Leu Ser Phe Arg Arg Arg Thr Asp
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 Lys Asp Thr Glu Gln Pro Gly Glu Val Ser Ala Leu Gly Pro Gly Arg
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 930 935 940
 Gly Pro Gly Arg Ser Ser Ser Pro Leu Arg Leu Val Pro Phe Ser Ser
 945 950 955 960
 Pro Arg Pro Pro Gly Glu Pro Pro Gly Gly Glu Pro Leu Met Glu Asp
 965 970 975

Cys Glu Lys Ser Ser Asp Thr Cys Asn Pro Leu Ser Gly Ala Phe Ser
 980 985 990
 Gly Val Ser Asn Ile Phe Ser Phe Trp Gly Asp Ser Arg Gly Arg Gln
 995 1000 1005
 Tyr Gln Glu Leu Pro Arg Cys Pro Ala Pro Thr Pro Ser Leu Leu Asn
 1010 1015 1020
 Ile Pro Leu Ser Ser Pro Gly Arg Arg Pro Arg Gly Asp Val Glu Ser
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 Ser Ala Asp Met Ala Thr Val Leu Gln Leu Leu Gln Arg Gln Met Thr
 1060 1065 1070
 Leu Val Pro Pro Ala Tyr Ser Ala Val Thr Thr Pro Gly Pro Gly Pro
 1075 1080 1085
 Thr Ser Thr Ser Pro Leu Leu Pro Val Ser Pro Leu Pro Thr Leu Thr
 1090 1095 1100
 Leu Asp Ser Leu Ser Gln Val Ser Gln Phe Met Ala Cys Glu Glu Leu
 105 1110 1115 1120
 Pro Pro Gly Ala Pro Glu Leu Pro Gln Glu Gly Pro Thr Arg Arg Leu
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 His Gly Ser Asp Pro Gly Ser
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 <213> Artificial Sequence

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 <223> Description of Artificial Sequence:Hypothetical
 sequence for the example of calculating homology.

<400> 5
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 tgc 63

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 <212> DNA
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<220>
 <223> Description of Artificial Sequence:Hypothetical
 sequence for example of calculating homology.

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 <211> 19
 <212> DNA
 <213> Homo sapiens

<400> 13
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<210> 14
 <211> 22
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<400> 14
 gcctgggcg cccctccatc aa

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<210> 15
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 <212> DNA
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<400> 15
 cacctcctcg ttggcattga c

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<210> 16
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<400> 16
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25

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<400> 17
 tacaccacct gcctccttgc tga

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<400> 18
 gccgcgccgt actctgagta g

21

<210> 19
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 <212> DNA
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<400> 19
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23

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19

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22

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<210> 27
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<400> 29
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<400> 41
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<400> 45
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<400> 46
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<210> 47
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<400> 55
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<400> 56
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<400> 57
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<210> 70
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<400> 70
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20

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<400> 71
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22

<210> 72
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<400> 72
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<210> 73
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<400> 73
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<210> 74
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<400> 74
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21

<210> 75
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22

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<400> 76
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22

<210> 77
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<400> 77
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21

<210> 78
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<400> 78
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<210> 79
 <211> 21
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<400> 79
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<400> 80
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<400> 81
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21

<210> 82
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<400> 83
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21

<400> 84
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<400> 85
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19

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21

<400> 89
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09-06-2008 17:00